

Using NCBI Medical Genetics Resources: MedGen, ClinVar, GTR

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Materials: <https://go.usa.gov/xVvYm>



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Outline

- Medical Genetics Resources
 - MedGen
 - ClinVar
 - Genetics Testing Registry (GTR)
 - Others
- Demonstration
 - Find records for a specified list of symptoms or clinical features
 - Find tests relevant to a clinical feature, gene or disease
 - Explore specific disease-causing variants
- Search tips

NCBI's Medical Genetics Resources

MedGen – phenotypes (diseases or conditions) and vocabularies

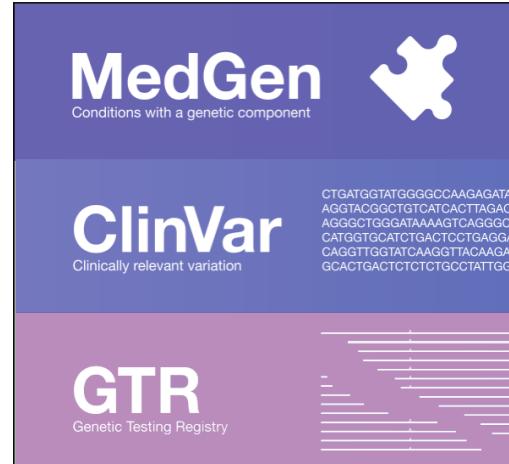
www.ncbi.nlm.nih.gov/medgen

ClinVar – submitted genetic variants and their relationships to disease

www.ncbi.nlm.nih.gov/clinvar

Genetic Testing Registry (GTR) – submitted genetic tests from testing laboratories

www.ncbi.nlm.nih.gov/gtr



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MedGen: Medical Genetics Terms and Conditions

- Terms and vocabularies from several sources
 - Genetic Testing Registry
 - Unified Medical Language System (UMLS)
 - Human Phenotype Ontology (HPO)
 - Ophanet
 - ClinVar
- Other data sources
 - Online Mendelian Inheritance in Man (OMIM)
 - GeneReviews
 - Genetics Home Reference
- Organized by concept ID with synonyms, cross-references and links to sources
- PubMed references (computed and curated)
 - Professional Guidelines
 - Recent Clinical Studies (Clinical Queries www.ncbi.nlm.nih.gov/pubmed/clinical)
 - Recent Systemic Reviews

MedGen Flyer: go.usa.gov/xVySu

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ClinVar: Clinical Variations

- ClinVar provides the primary submission route for clinically relevant human genetic variants ClinVar Flyer: go.usa.gov/xVyha
- Submitted genetic variants with clinical assertions (phenotypes)
- Large-scale (>50 bp to millions of bp) and small-scale (<50 bp variants)
 - Primary data in **dbVar** for large-scale and in **dbSNP** for small-scale variants
- Sources include literature (OMIM), genetic testing consortia and individual labs

ClinVar Data (SCV, VCV, RCV)

- The submitted variant with a phenotype (interpretation, condition) is the basic unit (SCV accession)
- We aggregate these data by
 - Variant (VCV accession)
 - Default web page view
 - Condition (RCV accession)
 - Conditions tab on ClinVar record
- The same variant may have a different interpretation for different conditions

Genetic Testing Registry (GTR)

- NIH's international registry of genetic tests
- Voluntary submissions from testing labs
- Intended for clinicians
- Searchable by test, conditions, genes, and labs
- Tests filterable by purpose, method, lab certification, lab location, and other aspects
- Specific tests details include ordering info, indication, methodology, performance, interpretation, contact info

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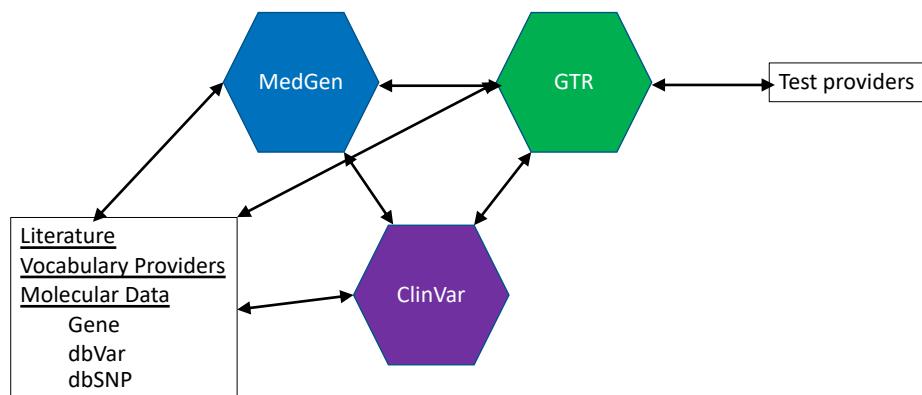
Other Medical Genetics Resources

- On the NCBI Bookshelf
 - GeneReviews® www.ncbi.nlm.nih.gov/books/NBK1116
 - Medical Genetics Summaries www.ncbi.nlm.nih.gov/books/NBK61999
- OMIM® omim.org
 - Flyer GeneReviews: go.usa.gov/xVyhB
 - Flyer Medical Genetics Summaries: go.usa.gov/xVyhY
- Consumer / Non-expert
 - Genetics Home Reference ghr.nlm.nih.gov
 - MedlinePlus® medlineplus.gov
- ClinicalTrials.gov clinicaltrials.gov
- Variation Resources www.ncbi.nlm.nih.gov/variation/

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Integrated Medical Genetics Resources



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Live Web Demonstration

Chronic Lung Disease-> Cystic Fibrosis ->del508 mutation and test

- Find records for a specified list of symptoms or clinical features (MedGen)
- Find tests relevant to a clinical feature, gene or disease (GTR)
- Explore specific disease-causing variants (ClinVar)

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A note on the deltaF508 CFTR variant

Reference Sequence		HGVS name for the change
1513	1520	NM_000492.3:c.1520_1522delTCT
AAT-AAT-AT C-T T-GGT-GTT		
Asn-Ile-Ile- Phe -Gly-Val		
1513	1520	Deletion of the TCT at 1520 or the CTT at 1521 gives the same result!
AAT-AAT-AT C-TT T-GGT-GTT		AAT-AAT-ATT-GGT-GTT
Asn-Ile-Ile- Phe -Gly-Val		Asn-Ile-Ile-Gly-Val
Reference Sequence		HGVS name for the change
		NM_000492.3:c.1521_1523delCTT

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Search Tips

Term	Best Databases
Phenotype (disease, condition)	MedGen, (GTR, ClinVar)
Specific Variant (HGVS, Other variant names)	ClinVar
Gene symbols	MedGen, ClinVar, GTR

- URLs & Places to Learn More -

- YouTube www.youtube.com/ncbinlm
 - ClinVar playlist bit.ly/2vT9ncu
 - GTR playlist bit.ly/2y2ihED
 - Medical Genetics Summaries youtu.be/4Mhzv8LQTa0
- Factsheets: <ftp.ncbi.nlm.nih.gov/pub/factsheets/>
 - MedGen go.usa.gov/xP94F
 - ClinVar go.usa.gov/xP92d
 - GTR go.usa.gov/xP92G
 - Medical Genetics Summaries <http://go.usa.gov/xVvSH>
- NCBI Listserves ncbi-announce@ncbi.nlm.nih.gov
 - Subscribe at www.ncbi.nlm.nih.gov/news/announcements-list/
- NCBI Insights Blog: ncbiinsights.ncbi.nlm.nih.gov
- Learn: www.ncbi.nlm.nih.gov/home/learn.shtml



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Questions?

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